

Special Issue

Molecular Effects of Mutations in Human Genetic Diseases

Message from the Guest Editors

The aim of this Special Issue is to attract high-quality studies describing computational and experimental approaches for investigating the molecular effects of novel genetic mutations, and providing a useful framework for understanding the molecular defects underlying human diseases. Contributors are also encouraged to submit articles describing use cases, models, and methodological innovations. The Special Issue will include (1) human genetics studies on genome/exome or targeted sequencing panels that allow either identification of disease–gene associations, characterization of rare diseases with significant genetic heterogeneity, or differential clustering of disease mutations associated with distinct phenotypes; (2) experimental studies investigating how a genetic variant causes disease at the molecular, cellular, and organismal levels; and (3) computational methods devised to predict the impact of genetic variations and their assessment, large-scale statistical studies dissecting key features of disease mutations, or well-curated data repositories of genetic variation and/or disease associations.

Guest Editors

Dr. Emanuela Leonardi

Dr. Giovanni Minervini

Dr. Castrense Savojardo

Deadline for manuscript submissions

closed (30 June 2021)



International Journal of Molecular Sciences

an Open Access Journal
by MDPI

Impact Factor 4.9
CiteScore 9.0
Indexed in PubMed



mdpi.com/si/48238

*International Journal of
Molecular Sciences*
Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
ijms@mdpi.com

[mdpi.com/journal/
ijms](https://mdpi.com/journal/ijms)





International Journal of Molecular Sciences

an Open Access Journal
by MDPI

Impact Factor 4.9
CiteScore 9.0
Indexed in PubMed



[mdpi.com/journal/
ijms](https://mdpi.com/journal/ijms)



About the Journal

Message from the Editor-in-Chief

The International Journal of Molecular Sciences (*IJMS*, ISSN 1422-0067) is an open access journal, which was established in 2000. The journal aims to provide a forum for scholarly research on a range of topics, including biochemistry, molecular and cell biology, molecular biophysics, molecular medicine, and all aspects of molecular research in chemistry. *IJMS* publishes both original research and review articles, and regularly publishes special issues to highlight advances at the cutting edge of research. We invite you to read recent articles published in *IJMS* and consider publishing your next paper with us.

Editor-in-Chief

Prof. Dr. Maurizio Battino

Department of Odontostomatologic and Specialized Clinical Sciences,
Sez-Biochimica, Faculty of Medicine, Università Politecnica delle
Marche, Via Ranieri 65, 60100 Ancona, Italy

Author Benefits

Open Access:

free for readers, with article processing charges (APC) paid by authors or their institutions.

High Visibility:

indexed within Scopus, SCIE (Web of Science), PubMed, PMC, MEDLINE, Embase, CAPlus / SciFinder, and other databases.

Journal Rank:

JCR - Q1 (Biochemistry and Molecular Biology) / CiteScore
- Q1 (Organic Chemistry)